Cystic Fibrosis (CF) Summary

Primary Defect

A defective protein disrupts the movement of salt and water across cell membranes. A primary effect of this is accumulation of thick mucus that interferes with lung and digestive function.

Screening Test

Immunoassay to measure the pancreatic enzyme immunoreactive trypsin (IRT) followed by DNA analysis for those with elevated levels. 3% to 5% false negative rate. Positive predictive value is nearly 100% when two defective genes are found, 5-10% when only a single defective gene is found (based on data from Wisconsin with DNA testing for the common mutation only).

Etiology & Prevalence

Genetic autosomal recessive. A single mutation (delta F508) is most common in Northern Europeans and is associated with severe clinical outcomes. However, hundreds of other mutations have been identified, some with more mild disease states. There is significant clinical variability, however, even among those with identical genetic defects.

Occurrence (and genotype) is highly variable among different populations. About 1 in 2000 Northern European, 1 in 17,000 African American and 1 in 9,000 Hispanic infants are affected.

If Untreated

Substantial impact on lung function, increased lung infections, and malnutrition due to abnormal production of pancreatic enzymes. Other features include cirrhosis of the liver, abnormal glucose tolerance, and infertility.

Therapy

Currently only palliative therapies are available. These are aimed primarily at maintaining lung function, preventing infection, and enhancing nutritional status.

With Treatment

Improved treatments have dramatically increased life expectancy for affected individuals from approximately 3 years in the 1950's to the late 30's today. The benefits of early detection through newborn screening are still not fully understood, however, research has clearly shown that there is long term clinical benefit when therapeutic interventions are administered near the beginning of life. A recent, careful review of the evidence has led the Centers for Disease Control and Prevention to conclude that newborn screening for cystic fibrosis is justified based on the scientific evidence of clinical utility.

GENERAL REFERENCES

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CF Personal story

We were unable to locate any personal stories on CF that focused on screening. The following is a mother's story describing her experience with her child who was born with meconium ileus which led to his early diagnosis. About 10% of infants with CF are born with this manifestation. There is also a Web site, **http://www.thebreathingrooom.org/lg/index.html,** that presents some very compelling photos and poems by persons living with CF.

Mother

I had no reason to believe that I would not have a perfectly healthy baby when I was pregnant with my son Joe. I was healthy, my husband was healthy, so why wouldn't our child be healthy, too? Soon after he was born in 1992, it became apparent that he was not only unhealthy, but had something major going on. He had not passed any meconium and had no interest in eating. An x-ray detected an obstruction and required immediate surgery. When he was one day old, Joe was airlifted to Children's Hospital in Seattle for the procedure. When he came out of surgery, our once perfect baby was penetrated by tubes and had an incision the width of his tiny belly. I couldn't hold him, I couldn't nurse him - I could only stare at him and watch the monitors moving and beeping to indicate he was OK, for now.

We were able to bring Joe home after a week in the hospital. No doctors, no nurses... we were now in charge of this little guy. The doctors mentioned the possibility of a disease called cystic fibrosis and asked if there were any family history on either side. To our knowledge, there wasn't. Since Joe was too small to take any necessary CF tests, doctors ran other tests in search of answers. At three months, he tested positive for CF. My husband and I then started to read all we could about CF - arming ourselves with knowledge was our only hope.

Joe is now a very active nine-year-old boy. He missed 63 days of school last year due to hospital stays and various infections. He spends an hour in the morning on his 'routine,' which includes a ½ hour with his 'vibrating vest,' two different nebulized medications and one inhaled medicine. His breakfast consists of buttery rice and steak served in a 'Jethro' sized bowl. Along with this, Joe has a handful of pills, which he is proud to tell you he can take four at a time. Kids with CF need extra fat and calories in their daily meals so a cold cereal or a Pop Tart won't do. He remains active in sports and enjoys Nintendo and cartoons - so in many ways he is just a 'normal' kid. Joe has friends, Joe has parents, Joe has two dogs, one cat, one fish - and Joe has cystic fibrosis.